

Findings: The results showed that although the mean of indexes in case group significantly lower than the control group, but all hypothyroidism children had ASQ score level higher than -1SD in aspect communication-motor-social-and fine course-solving.

Conclusion: Although the average indices evolve based on ASQ in hypothyroid children that treated was less than the control group but was acceptable.

Keywords: Hypothyroidy, Development of Children, ASQ Questionnaire

Molecular diagnosis of PKU: application of Next Generation Sequencing

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PKU is a common and highly heterogeneous metabolic disorder. The incidence of this disorder has been estimated at 1 in 6000-8000 in Iranian population. This inborn error of metabolism (IEM) mostly characterized by a deficiency of the phenylalanine hydroxylase (PAH) and rarity is due to tetra hydrobiopterin (BH4) cofactor deficiency. A striking feature of the disease is its genetic heterogeneity, best illustrated by the fact that 750 different mutations in PAH gene have been identified yet. Identifying the genetic cause of the patients' disease is crucial for genetic counseling, and is a prerequisite for any form of genotype-based therapies. The direct approach for diagnosis essentially depends on the detection of the genetic variations responsible for the disease. However, the enormous genetic heterogeneity in PAH makes attempts to identify causative mutations a challenging task. Next-generation sequencing (NGS) systems provide several sequencing approaches including whole genome sequencing and whole exome sequencing. This technology is capable of sequencing all PKU-associated gene mutations in parallel, generating millions of reads from preselected genomic regions. Ongoing cost reduction and the development of standardized pipelines will probably make NGS a standard tool for more-routine applications in the near future. Indeed, NGS technologies bring us new sights in unraveling the genetic basis of diseases. Not surprisingly, as the technology continues to improve, NGS-based tests may become stand-alone, without the need for confirmation through a second method.

Keywords: Phenylketonuria, Next Generation Sequencing, genetic counseling

Prevalence of diabetic ketoacidosis in new cases of type I diabetes admitted to the children hospital in Qazvin, Iran (2005-2014)

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Background: Type 1 diabetes (T1D) is one of the most common chronic diseases in childhood and adolescence. Diabetic ketoacidosis (DKA) is a severe complication of T1D and is associated with significant morbidity and mortality. The aim of this study was to determine the prevalence of DKA in new cases of T1D admitted to the children hospital in Qazvin during 2005-2014. **Methods:** In this cross sectional study, data were collected from health profiles of 115 patients that were hospitalized as new cases of T1D during 2005-2014. The measurement tool was a datasheet including demographics, signs and symptoms of T1D, characteristics at the onset of disease, and characteristics during the hospitalization period. DKA was defined as blood glucose ≥ 250 mg/dl, arterial PH < 7.3, serum bicarbonate (HCO_3) > 15 meq/L, and positive urine ketones. Data were analyzed using descriptive statistics. **Findings:** Of 115 patients, 62. 6% were female. The onset of T1D was diagnosed by DKA and hyperglycemia in 94. 7% and 5. 3% of patients, respectively. 76 (71. 1%) of the study subjects had severe DKA and only one subject had mild DKA. Twenty seven percent of patients were

Keywords: Diabetic Ketoacidosis, Child, Type 1 Diabetes Mellitus

Ovarian Hyperstimulation Syndrome in Preterm Infants

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Background: Ovarian Hyperstimulation Syndrome in Preterm Infants Ovarian hyperstimulation is a rare syndrome among preterm infants. The patients show ovarian cysts and external genitalia edema extending to hypogastric and upper leg regions. The hormonal changes in this syndrome include different degrees of increased gonadotropin and estradiol levels.